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Case Report

Rhombencephalosynapsis: A Rare Cerebellar Malformation

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Abstract

Background: Rhombencephalosynapsis is characterized by agenesis/hypogenesis of the cerebellar vermis along with the fusion of the cerebellar hemispheres in the midline. There are less than 50 cases in the literature, and adult cases are extremely rare. In this paper, an adult patient with rhombencephalosynapsis is reported.
Case: A 24-year-old female was admitted with the complaint of headache. Neurological examination was completely normal, and past medical history was eventless. Magnetic resonance imaging revealed agenesis of the vermis along with the fusion of the cerebellar hemispheres.
Conclusion: Although a rare malformation, rhombencephalosynapsis should be considered in the differential diagnosis of the cerebellar malformations.

Keywords: Cerebellum, magnetic resonance imaging, rhombencephalosynapsis

Rombensefalosinapsis: Nadir Bir Serebellar Malformasyon

Özet

Amaç: Rombensefalosinapsis, serebellar vermisin agenezisi/hipogenezisi ve serebellar hemisferlerlerin birbirine kaynaması ile karakterize bir malformasyondur. Bugüne kadar elliden az olgu bildirilmiş olup, erişkin olgular son derece nadirdir. Bu makalede, erişkin bir rombensefalosinapsis olgusu bildirilmektedir.
Olgu: 24 yaşında kadın hasta, kliniğimize başağrısı şikâyeti ile başvurdu. Yapılan nörolojik muayenede patoloji saptanmadı. Hastanın özgeçmişinde özellik yoktu. Yapılan manyetik rezonans görüntüleme tetkikinde vermisin olmadığı ve serebellar hemisferlerin ortahatta birbirine kaynamış olduğu görüldü.
Sonuç: Nadir olarak görülse de, rombensefalosinapsis, serebellar malformasyonların ayırıcı tanısında mutlaka akılda bulundurulmalıdır.

Anahtar Kelimeler: Manyetik rezonans görüntüleme, rombensefalosinapsis, serebellum

INTRODUCTION

Rhombencephalosynapsis is characterized by agenesis/hypogenesis of the cerebellar vermis along with the fusion of the cerebellar hemispheres in the midline [6]. Less than 50 cases have been reported in the literature, and great majority of these cases belongs to pediatric age group. Newly diagnosed cases in the adulthood are extremely rare [1]. In this paper, an adult patient with rhombencephalosynapsis is reported.

CASE PRESENTATION

A 24-year-old female was admitted to our neurosurgery out-patient clinic, with the complaint of headache. It was non-focal, squeezing type, not associated with nausea, vomiting or visual disturbances. The patient

was also complaining about her problems at work and at home. General examination and neurological examination were completely normal. Her past medical history was eventless. A cranial magnetic resonance imaging (MRI) was ordered at patient's request. On MRI, there was agenesis of the vermis along with the fusion of the cerebellar hemispheres (Fig 1). A radiological diagnosis of rhombencephalosynapsis was made. The clinical picture was not attributed to this diagnosis, but rather considered as a tension-type headache. A tricyclic antidepressant was initiated along with a non-steroidal anti inflammatory drug, and follow-up examinations were planned.

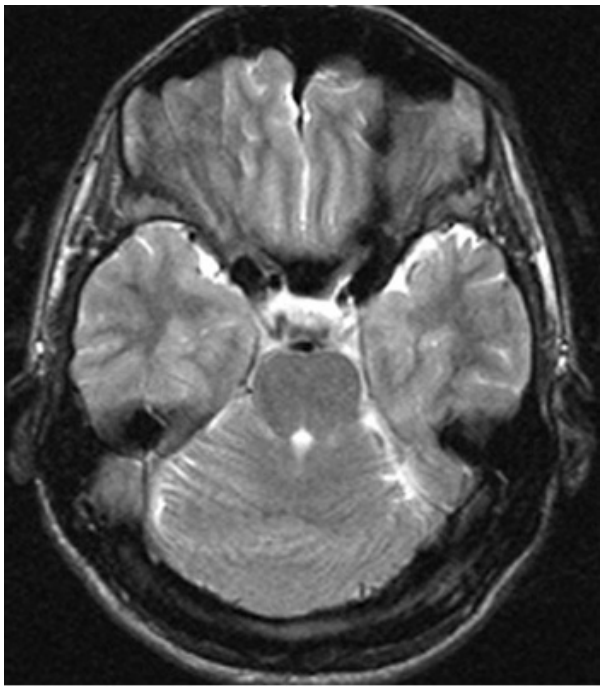

Figure 1: *T2-weighted axial MRI scan of the patient.*

DISSCUSSION

Rhombencephalosynapsis is characterized by agenesis/hypogenesis of the cerebellar vermis along with the fusion of the cerebellar hemispheres in the midline [6]. First case in the literature was described by Obersteiner in 1914, in a post-mortem study [3]. Since then, less than 50 cases have been reported, and great majority of these cases belongs to pediatric age group. Newly diagnosed cases in the adulthood are extremely rare [1].

Rhombencephalosynapsis is a vermian maldevelopment syndrome, but unlike other vermian maldevelopment syndromes, such as Dandy-Walker malformation or Joubert syndrome, there is no disconnection of the cerebellar hemispheres [4]. On the contrary, cerebellar hemispheres are fused, and they continue seamlessly through the midline. As a matter of fact, fusion is not the right term, from the scope of embryology, because cerebellar primordium is an uncoupled structure, and failure of vermian development leads to undivided cerebellar hemispheres, instead of fused ones [5]. Another differentiating factor is, in Dandy-Walker malformation posterior vermis tends to be defective, whereas in rhombencephalosynapsis, anterior vermis is absent and posterior vermis tends to be defective [8].

No specific clinical picture has been associated with rhombencephalosynapsis [6]. Largest series in the literature consists of 9 pediatric patients [7]. In these patients clinical picture was reported to be varying from mild truncal ataxia and normal cognitive abilities to severe cerebral palsy and mental retardation. There was no correlation between the neuroimaging findings and the clinical manifestations. Although behavioral and intellectual impairment is generally reported in rhombencephalosynapsis, it's not inevitably associated [1].

Nineteen cases have been reported in the pre-MRI era, and all the cases were detected at necropsy [8]. Rest of the cases was diagnosed by MRI. There is no case in the literature, which is diagnosed solely on computed tomography (CT) findings [2]. Associated findings on MRI include; deficiency or absence of the septum pellucidum, dysgenesis of the corpus callosum and the anterior commisure, fused fornices and thalami [8]. Superior and middle cerebellar peduncles, along with dentate nuclei and inferior colluculi, are generally fused, giving rise to the typical diamond-shaped fourth ventricle on axial scans. Aqueductal stenosis is commonly associated, and it represents as congenital hydrocephalus [9].

As a conclusion, although a rare malformation, rhombencephalosynapsis should be considered in the differential diagnosis of the cerebellar malformations.

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